Mutation sequence analysis			
Contributed by : CHU Lille			
HGVS nomenclature (NM_000295.4)			
Nomenclarure including the signal peptide			
c.250G>A			
Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 2	M1	Uncertain significance
Comments			
rs111850950			
AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$M_{6passau}$		Unknown	p.Ala84Thr

3D position of aa affected	Mobility on polyacrylamide gel	Mobility on agarose gel
		M

AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	**Homozygous**	**Heterozygous**	**Homozygous**
1.17		18290	

Comments
associated with a M1 allele.
Occurrence
Ethnic background without frequency range :
Ethnic background and frequency

Frequency range		**Group tested**	
from (%)	To (%)	Size	Description (who was tested)
0.02	0.06		

Occurrence comments

from gnomAD (2.1)

Overall comments

Occurrence comments

This variant was identified at a heterozygous status in a 45-year old man presenting with bronchiectasis and a severe pulmonary obstruction.

References n°1

Medline ID	**Authors**	**Title**
27296815	Silva D,Oliveira MJ,Guimarães M,Lima R,Gomes S,Seixas S	Alpha-1-antitrypsin (SERPINA1) mutation spectrum: Three novel variants and haplotype characterization of rare deficiency alleles identified in Portugal.

Journal	**Year**	**Volume**	**Num**	**Pp**
Respiratory medicine	2016	116		8-18

References n°2

Medline ID	**Authors**	**Title**
7977369	Faber JP,Poller W,Weidinger S,Kirchgesser M,Schwaab R,Bidlingmaier F,Olek K	Identification and DNA sequence analysis of 15 new alpha 1-antitrypsin variants, including two PI*Q0 alleles and one deficient PI*M allele.

Journal	**Year**	**Volume**	**Num**	**Pp**
American journal of human genetics	1994	55	6	1113-21

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